The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

FAQ

FREQUENTLY ASKED QUESTIONS
FAQ164
PREGNANCY

Prenatal Genetic Diagnostic Tests

What is prenatal genetic testing?

Prenatal genetic testing gives parents-to-be information about whether their ***fetus*** has certain ***genetic disorders***.

What are genetic disorders?

Genetic disorders are caused by changes in a person's ***genes*** or ***chromosomes***. ***Aneuploidy*** is a condition in which there are missing or extra chromosomes. In a ***trisomy***, there is an extra chromosome. In a ***monosomy***, a chromosome is missing. Inherited disorders are caused by changes in genes called ***mutations***. Inherited disorders include ***sickle cell disease***, ***cystic fibrosis***, ***Tay–Sachs disease***, and many others. In most cases, both parents must carry the same gene to have an affected child.

What are the two main types of prenatal genetic tests?

There are two general types of prenatal tests for genetic disorders:

1. Prenatal ***screening tests***: These tests can tell you the chances that your fetus has an aneuploidy and a few additional disorders. FAQ165 "Prenatal Genetic Screening Tests" focuses on these tests.
2. Prenatal ***diagnostic tests***: These tests can tell you, with as much certainty as possible, whether your fetus actually has an aneuploidy or specific inherited disorders for which you request testing. These tests are done on ***cells*** from the fetus or ***placenta*** obtained through ***amniocentesis*** or ***chorionic villus sampling (CVS)***. This FAQ focuses on these tests.

Both screening and diagnostic testing are offered to all pregnant women.

What is amniocentesis?

Amniocentesis is a diagnostic test. It usually is done between 15 weeks and 20 weeks of pregnancy, but it also can be done up until you give birth. To perform the test, a very thin needle is used to withdraw a small amount of ***amniotic fluid***. ***Ultrasound*** is used to guide the procedure. Depending on the way the cells are analyzed and the information that you want, results can take from 1 day to several weeks. There is a very small chance of pregnancy loss with amniocentesis. Leakage of amniotic fluid and slight bleeding can occur after amniocentesis. In most cases, both stop on their own.

What is chorionic villus sampling (CVS)?

CVS is another type of diagnostic test. In CVS, a sample of tissue is taken from the placenta. The two main advantages of having CVS over amniocentesis are that 1) CVS is performed earlier than amniocentesis, between 10 weeks and 13 weeks of pregnancy, and 2) the results are usually ready sooner for standard testing. With an experienced doctor, CVS carries about the same risk of pregnancy loss as amniocentesis.

What is preimplantation genetic diagnosis?

Preimplantation genetic diagnosis may be offered to couples who are using ***in vitro fertilization*** to become pregnant and who are at increased risk of having a baby with a genetic or chromosomal disorder. Before an ***embryo*** is transferred to a woman's uterus, it is tested for certain genetic disorders and mutations. Only embryos that do not test positive for the disorders are transferred.

How are the cells analyzed in prenatal diagnostic testing?

A number of technologies are used in prenatal diagnostic testing. Your ***obstetrician*** or ***genetic counselor*** can assess what information is being sought and help select the tests that are best for your situation:

- ***Karyotype***—Missing, extra, or damaged chromosomes can be detected by taking a picture of the chromosomes and arranging them in order from largest to smallest. Karyotyping results are ready in 1–2 weeks after the cells are sampled.
- ***Fluorescence in situ hybridization (FISH)***—This technique can be used to detect common aneuploidies involving chromosomes 13, 18, and 21 and the X and Y chromosomes. Results are ready more quickly (usually within 1–2 days) than with traditional karyotyping. Positive test results are confirmed with a karyotype.
- Chromosome ***microarray*** analysis—This test can look for different kinds of chromosome problems, including aneuploidy, throughout the entire set of chromosomes. It can find some chromosome problems that karyotyping can miss. Results can be ready in about 7 days.
- ***DNA*** testing—Tests for specific gene mutations can be done by request. For example, if you and your partner are ***carriers*** of the cystic fibrosis gene, you may want to request prenatal diagnostic testing for this specific mutation.

What do the different results of prenatal diagnostic tests mean?

Most of the time, the results of a diagnostic test are negative (normal). A negative result does not rule out the possibility that the fetus has a genetic disorder. It only tells you that the fetus does not have the particular disorder that was tested for.

If a diagnostic test result is positive (it shows that the fetus has the disorder tested for), your obstetrician or genetic counselor can explain the results and provide guidance about your choices and options. A specialist in the disorder can help you understand the life expectancy of the disorder, whether treatment is available, and the care that your child will need. Support groups, counselors, and social workers also can listen to your concerns and answer questions. It may be possible to have additional testing, such as a specialized ultrasound exam, to find out more detail about the defect.

What should I consider when deciding whether to have prenatal genetic testing?

It is your choice whether to have prenatal testing. Your personal beliefs and values are important factors in the decision about prenatal testing.

It can be helpful to think about what you would do if a diagnostic test result comes back positive. Some parents want to know beforehand if their child will be born with a genetic disorder. This gives parents time to learn about the disorder and plan for medical care that the child may need. If the disorder is very serious and the life expectancy is short, ***hospice care*** for the baby can be planned. Some parents may decide to end the pregnancy in certain situations. Other parents do not want to know this information before the child is born. They may decide not to have any testing at all. There is no right or wrong answer.

Keep in mind that certain tests can be done only at certain times during pregnancy. Tests that are done earlier allow parents more time to make decisions if a test result is positive. If ending the pregnancy is being considered, it is safer to do so within the first 13 weeks of pregnancy.

How do I choose between prenatal screening and diagnostic testing?

Any woman can choose to have diagnostic testing instead of or in addition to screening. The main benefit of having diagnostic testing instead of screening is that it can detect all conditions caused by an extra chromosome and many other disorders in which chromosomes are missing or damaged. Diagnostic tests also are available for many inherited disorders. The main disadvantage is that diagnostic testing carries a very small risk of losing the pregnancy. A genetic counselor or other health care professional with expertise in genetics can study your family health history, recommend specific tests, and interpret test results.

Glossary

Amniocentesis: A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

Amniotic Fluid: Water in the sac surrounding the fetus in the woman's uterus.

Aneuploidy: Having an abnormal number of chromosomes. Types include trisomy, in which there is an extra chromosome, or monosomy, in which a chromosome is missing. Aneuploidy can affect any chromosome, including the sex chromosomes.

Down syndrome (trisomy 21) is a common aneuploidy. Other trisomies include trisomy 13 (Patau syndrome) and trisomy 18 (Edwards syndrome).

Carriers: People who show no signs of a particular disorder but could pass the gene for the disorder on to their children.

Cells: The smallest units of a structure in the body; the building blocks for all parts of the body.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Chromosomes: Structures that are located inside each cell in the body and contain the genes that determine a person's physical makeup.

Cystic Fibrosis: An inherited disorder that causes problems in digestion and breathing.

Diagnostic Tests: Tests that look for a disease or cause of a disease.

DNA: The genetic material that is passed down from parents to offspring. DNA is packaged in structures called chromosomes.

Embryo: The stage of prenatal development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

Fetus: The stage of prenatal development that starts 8 weeks after fertilization and lasts until the end of pregnancy.

Fluorescence In Situ Hybridization (FISH): A laboratory technique that is used to screen for common chromosome problems, such as trisomy 21, in cells obtained by amniocentesis or chorionic villus sampling. Results are available fairly quickly because the cells do not need to be grown in a culture prior to testing.

Genes: Segments of DNA that contain instructions for the development of a person's physical traits and control of the processes in the body. They are the basic units of heredity and can be passed down from parent to offspring.

Genetic Counselor: A health care professional with special training in genetics and counseling who can provide expert advice about genetic disorders and prenatal testing.

Genetic Disorders: Disorders caused by a change in genes or chromosomes.

Hospice Care: Care that focuses on providing comfort rather than a cure for people who are in the final stages of a terminal illness.

In Vitro Fertilization: A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Karyotype: An image of a person's chromosomes, arranged in order of size.

Microarray: A technology that examines all of a person's genes to look for certain genetic disorders or abnormalities. Microarray technology can find very small genetic variations that have gone undetected by conventional genetic tests.

Monosomy: A condition in which there is a missing chromosome.

Mutations: Permanent changes in genes that can be passed on from parent to child.

Obstetrician: A physician who specializes in caring for women during pregnancy, labor, and the postpartum period.

Placenta: Tissue that provides nourishment to and takes waste away from the fetus.

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are performed on the fertilized egg before it is transferred to the uterus.

Screening Tests: Tests that look for possible signs of disease in people who do not have signs or symptoms.

Sickle Cell Disease: An inherited disorder in which red blood cells have a crescent shape, causing chronic anemia and episodes of pain. It occurs most often in African Americans.

Tay–Sachs Disease: An inherited birth defect that causes intellectual disability, blindness, seizures, and death, usually by age 5 years. It most commonly affects people of Eastern and Central European Jewish, Cajun, and French Canadian descent, but it can occur in anyone.

Trisomy: A condition in which there is an extra chromosome.

Ultrasound: A test in which sound waves are used to examine internal structures. During pregnancy, it can be used to examine the fetus.

If you have further questions, contact your obstetrician–gynecologist.

FAQ164: Designed as an aid to patients, this document sets forth current information and opinions related to women's health. The information does not dictate an exclusive course of treatment or procedure to be followed and should not be construed as excluding other acceptable methods of practice. Variations, taking into account the needs of the individual patient, resources, and limitations unique to the institution or type of practice, may be appropriate.